A RARE CAUSE OF SEIZURES

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CASE PRESENTATION

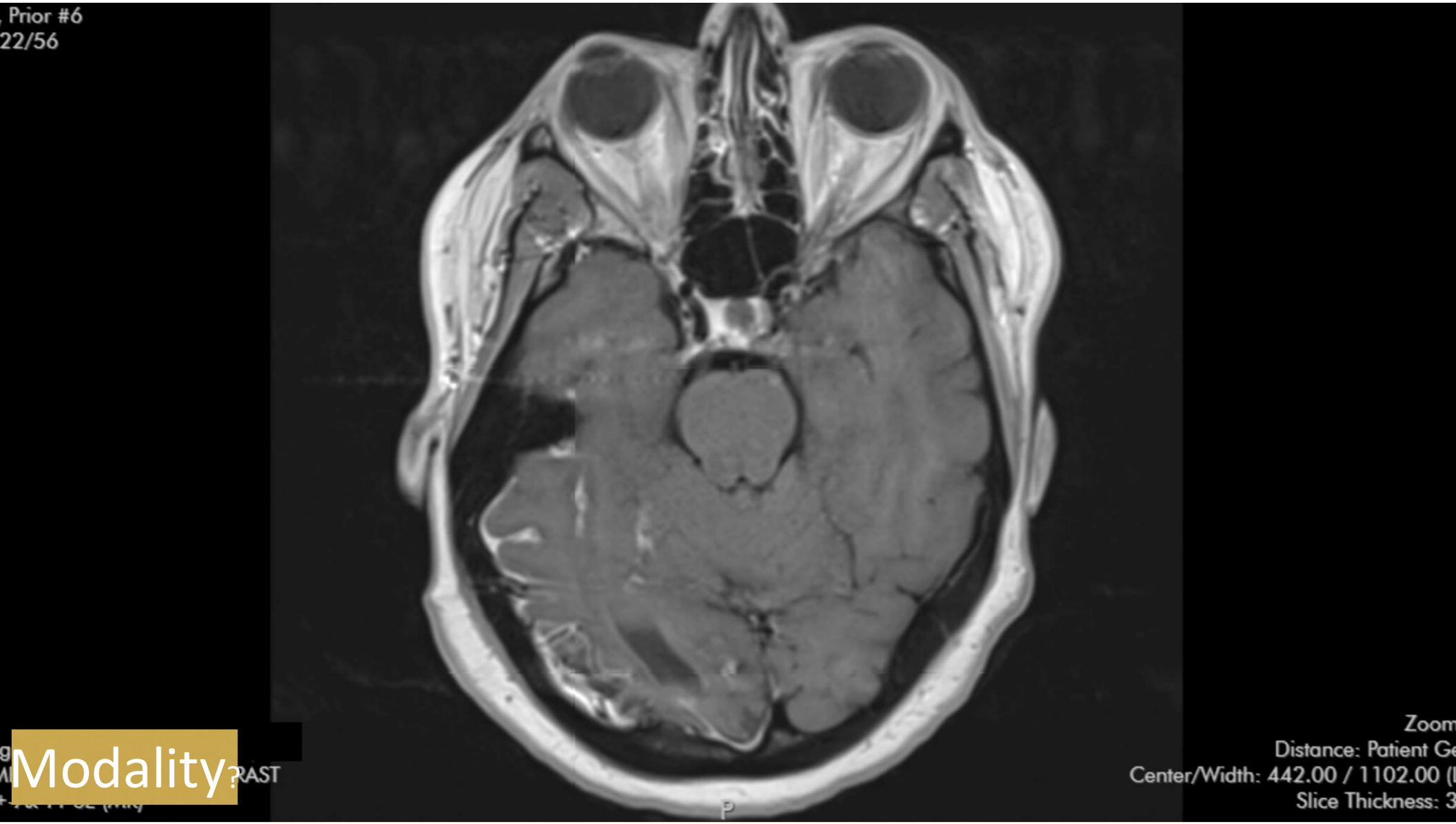
- 47 year old female presents to family medicine with several months of right arm weakness and visual disturbances. She has pain originating in her right elbow that radiates both proximally and distally along the upper extremity. There is associated numbness and tingling that extends from her elbow to hand in a diffuse distribution. She describes it as if something is “eating her muscle” and that there is “heat in her bones.” Her vision intermittently becomes blurry in her right eye but there is never complete loss of vision. She is very concerned about multiple sclerosis because her aunt was diagnosed with it. She was sent for MRI with contrast as outpatient.

HISTORY

- PMH - seizures in childhood, sleep apnea, migraine with aura, Crohn's disease, hyperlipidemia, and anxiety
- Family history
  - Mother - myocardial infarction
  - Father - testicular cancer (age 35), myocardial infarction, bilateral pulmonary embolism
  - Maternal aunt - multiple sclerosis and breast cancer

PHYSICAL

- Positive Phalen's test, Positive Finkelstein's sign, Negative Tinel sign
- Diffuse numbness over right hand and anterior forearm
- No visual field deficits, PERRLA, extraocular movements intact
- No rashes or erythema of skin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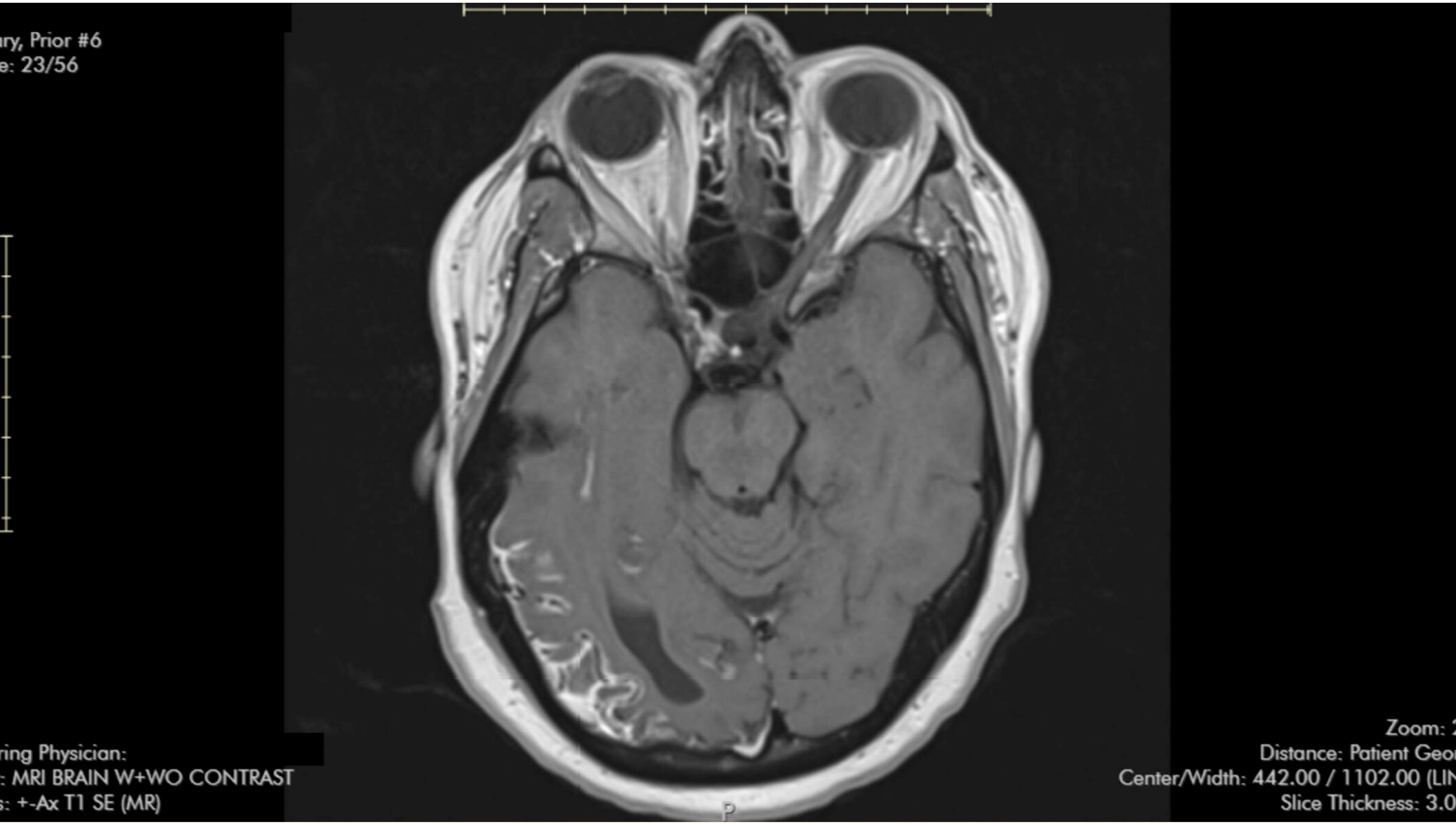
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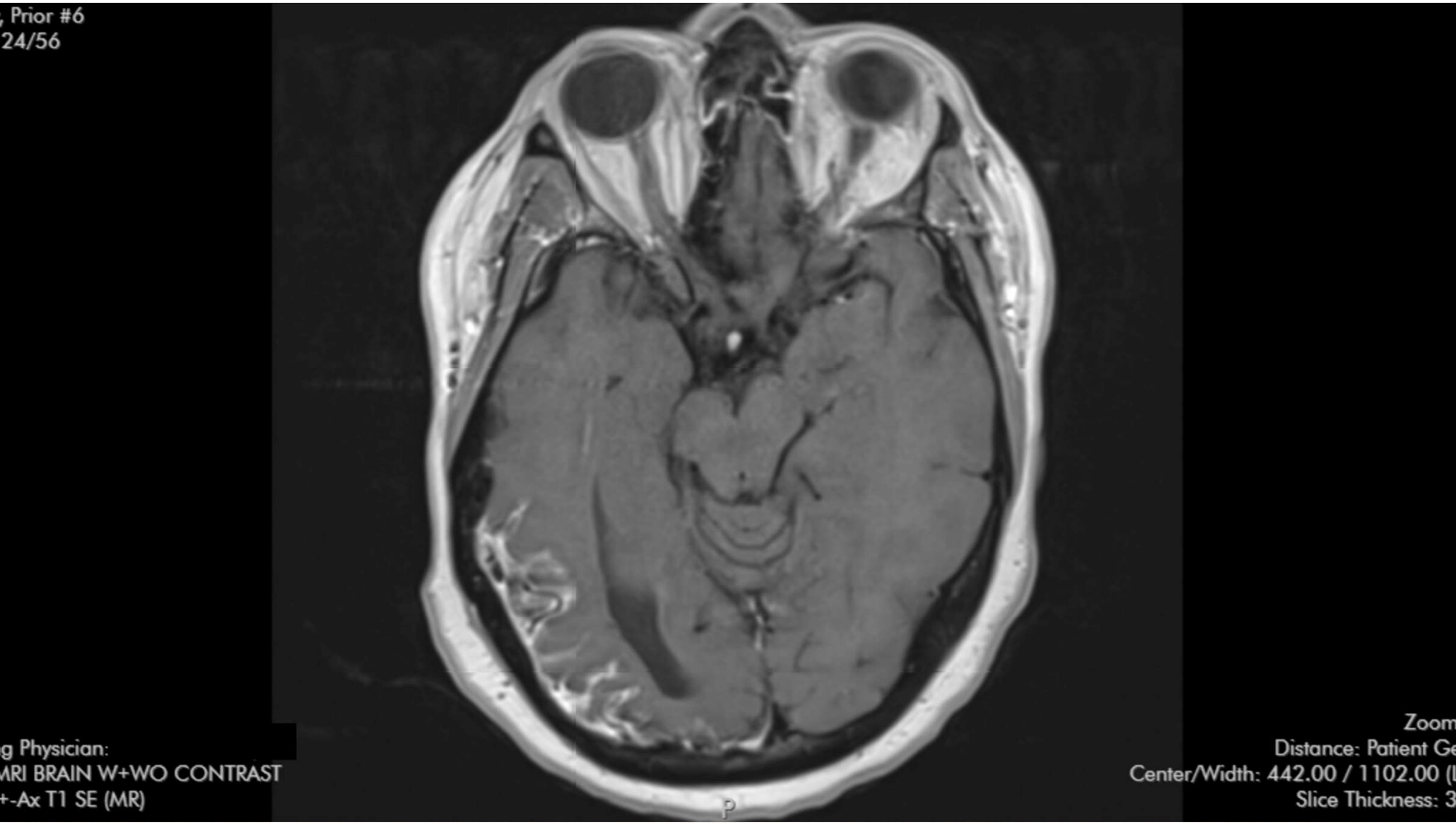
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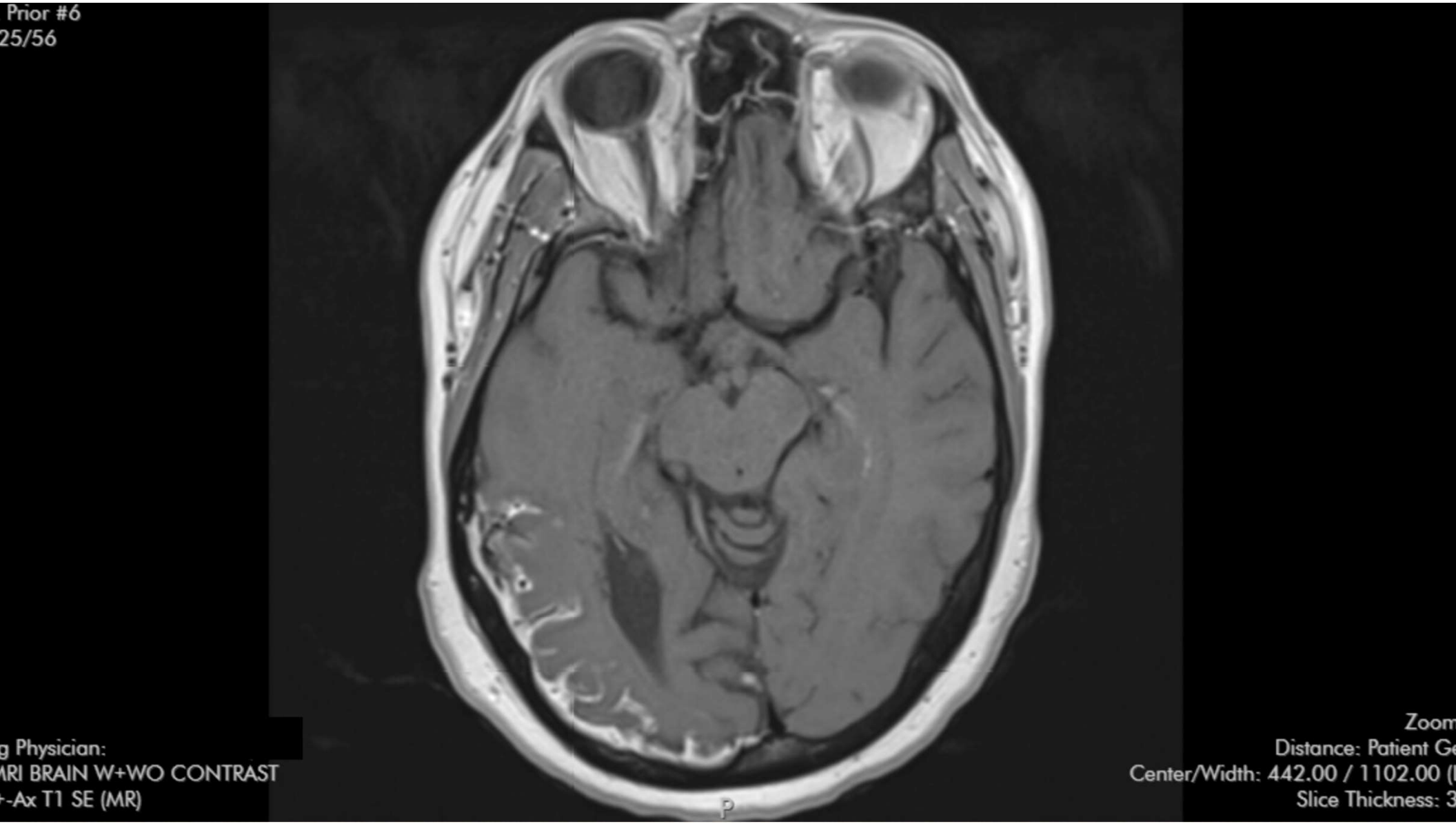
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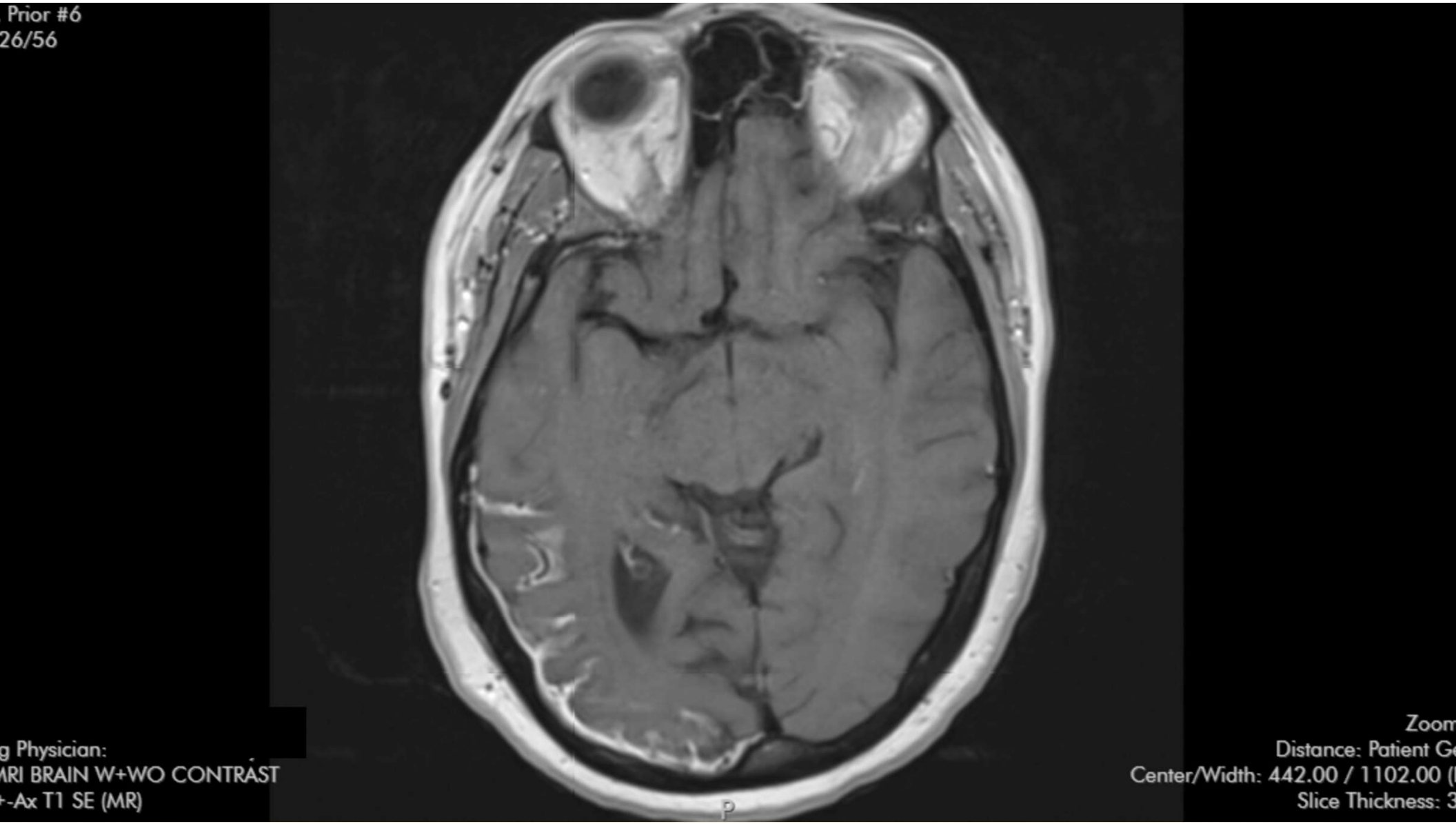
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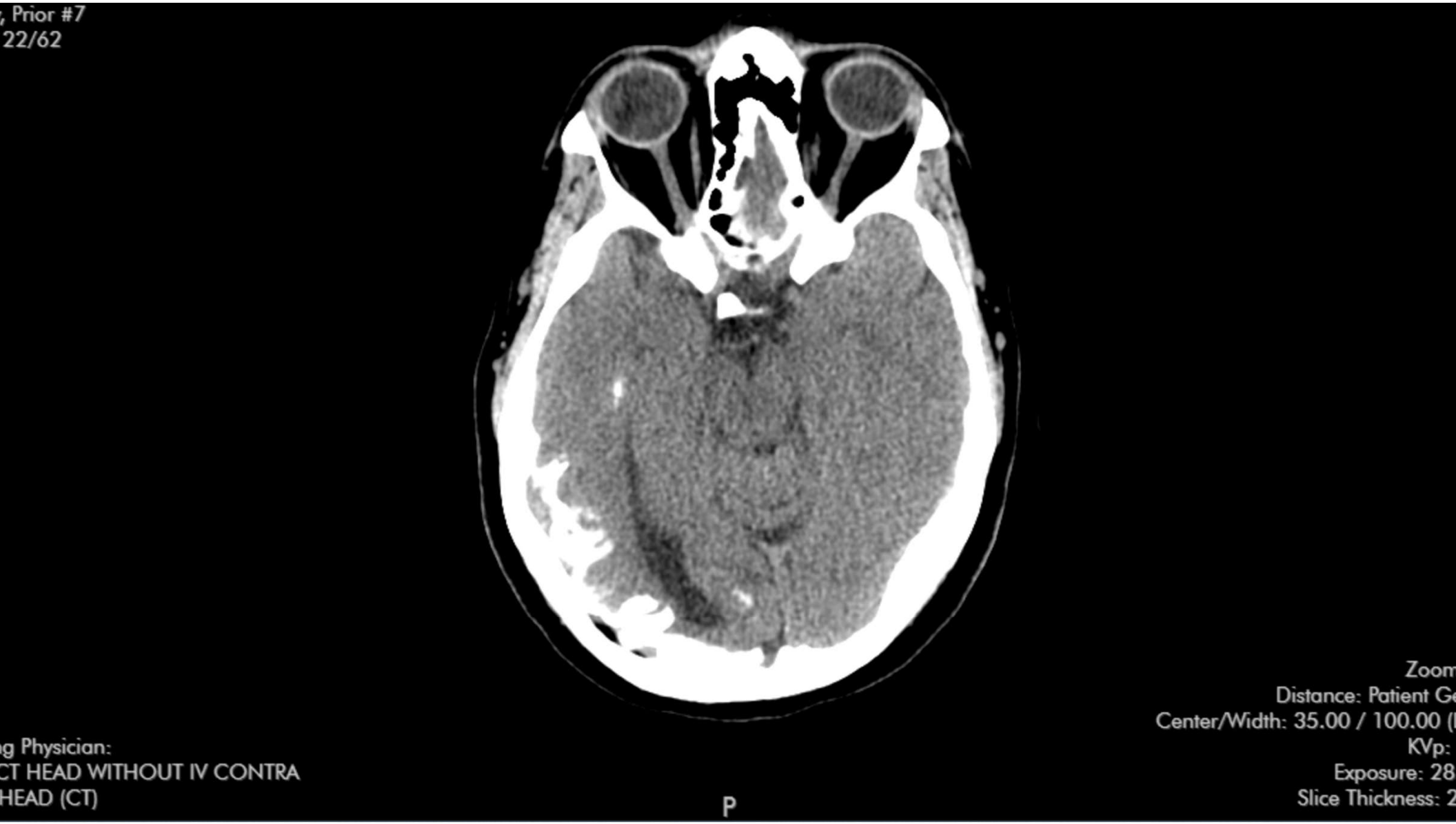
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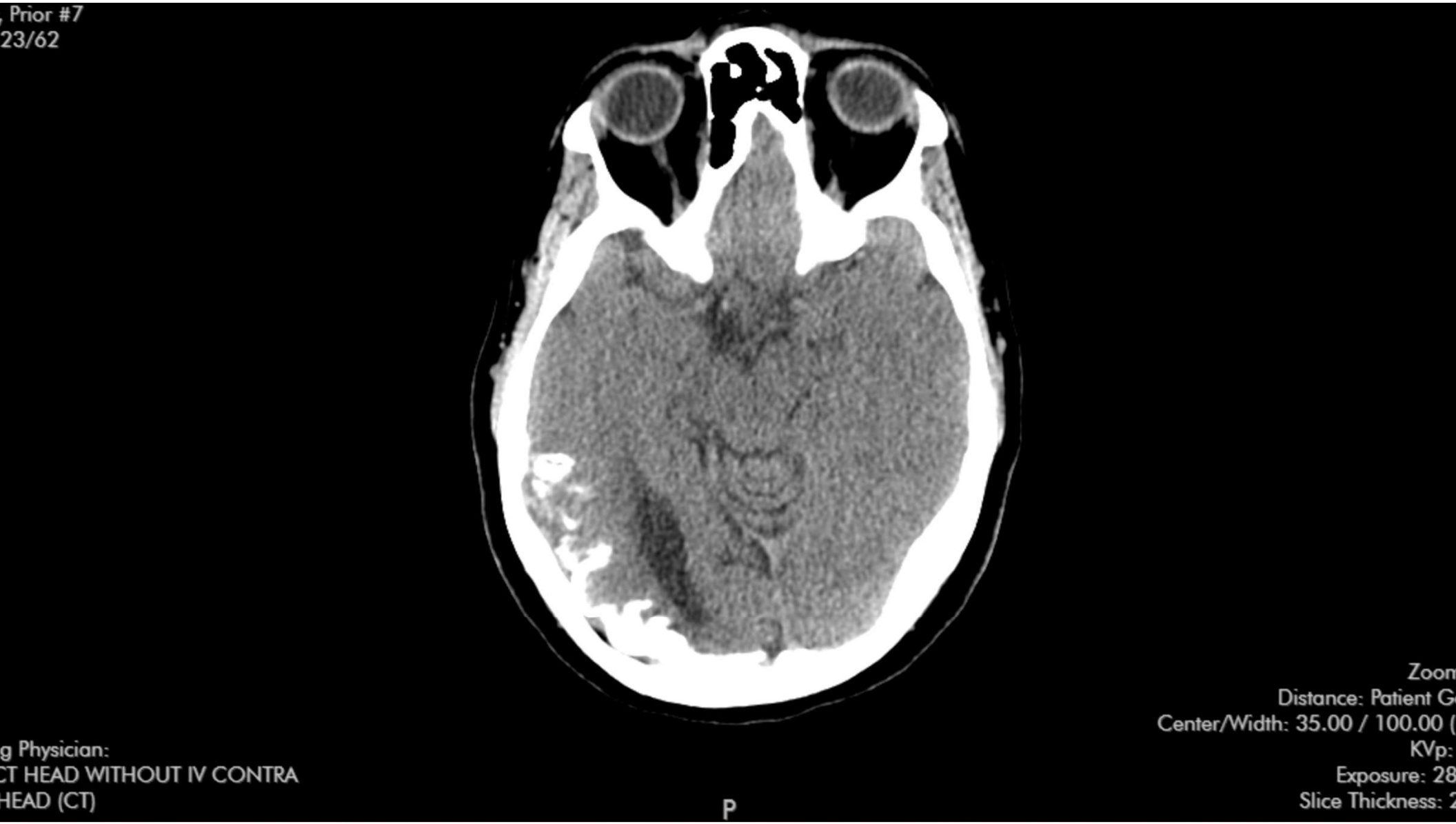
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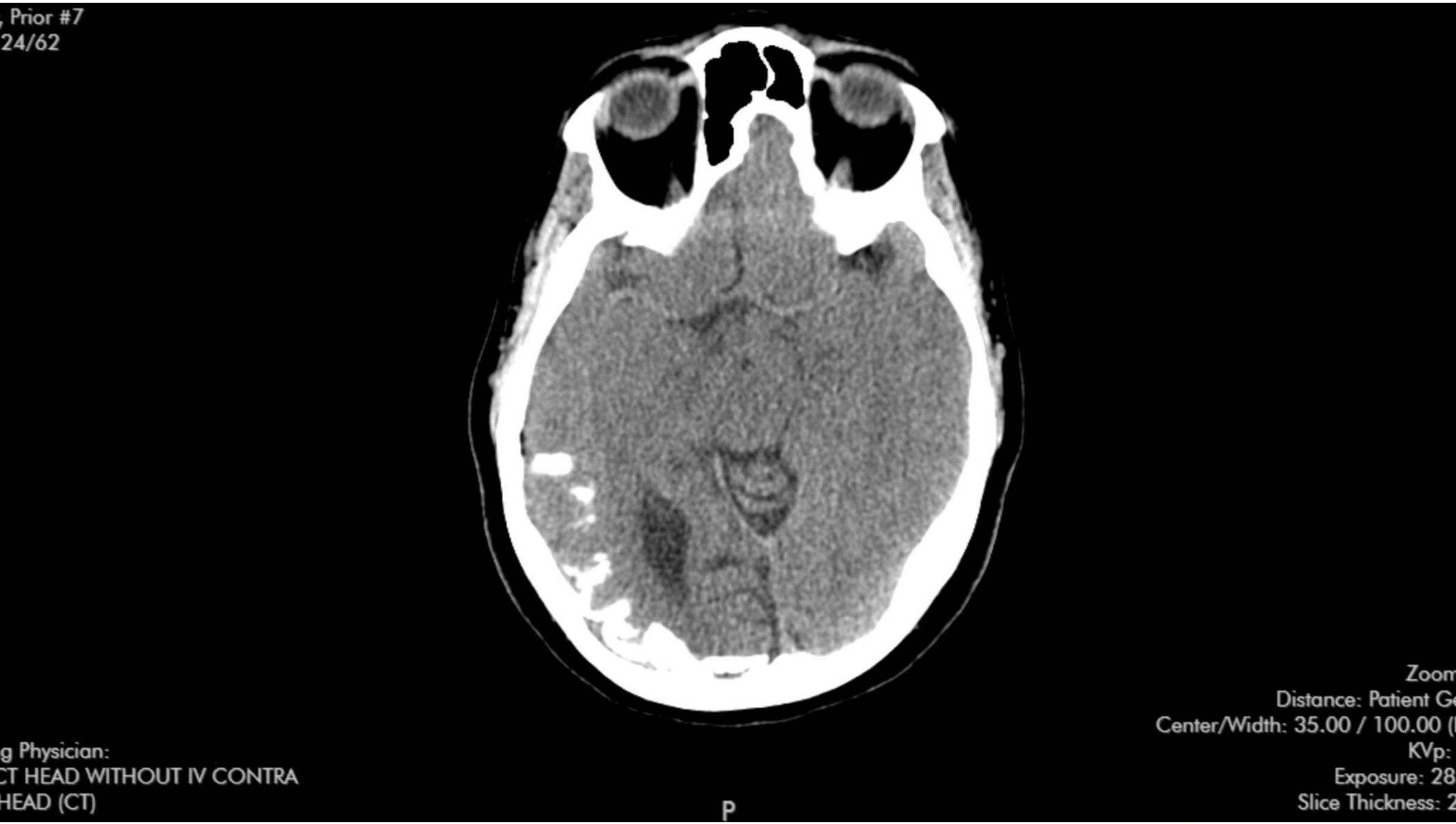
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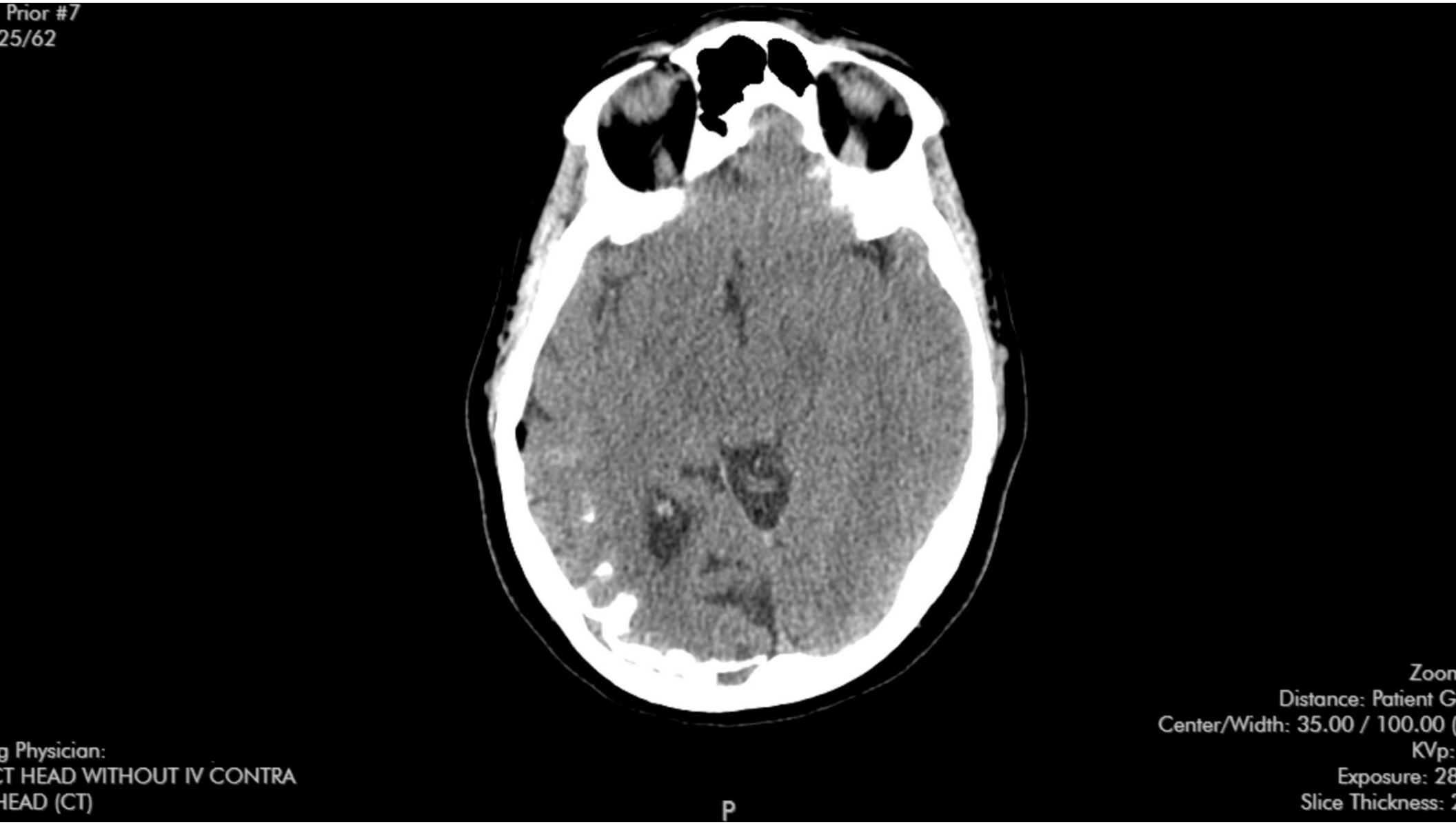
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DIFFERENTIAL DIAGNOSIS

- Sturge-Weber Syndrome
  - Somatic mutation classically causing a port wine stain, tram track gyral calcifications, intellectual disability, glaucoma, and epilepsy
- PHACE Syndrome
  - Posterior fossa malformations, Hemangiomas, Arterial anomalies, Coarctation of the aorta, and Eye anomalies
- Gobbi (CEC) Syndrome
  - Combination of celiac disease, epilepsy, and bilateral occipital calcifications
- Arteriovenous malformation

FINAL RADIOGRAPHIC REPORT

- There is evidence of leptomeningeal enhancement and abnormal signal intensity in the right posterior parieto-occipital region. These findings are compatible with patient's known changes of Sturge-Weber syndrome. Localized loss of cerebral volume and enlargement of the right occipital horn is also noted in this location.
- No significant change since last CT. No findings to suggest acute intracranial process.
- CT w/o contrast from two years prior - gyral cortical calcifications throughout right occipital and posterior temporal lobe with a slightly dysplastic brain and mild enlargement of the occipital horn of the right lateral ventricle.

MRI VS CT

- Advantages: sharper images, better visualization of soft tissue, no radiation exposure

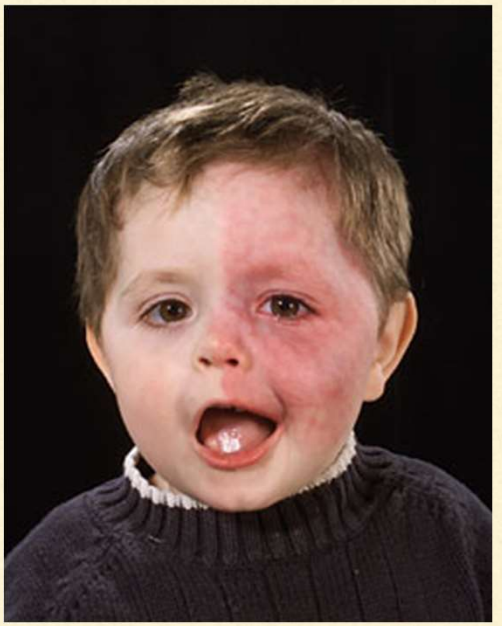
- Advantages: cheaper, faster, better visualization of bone, can use in patients with metal implants

STURGE-WEBER SYNDROME

- Epidemiology
  - 1 in 20,000-50,000 newborns
  - Third most common neurocutaneous disorder behind neurofibromatosis and tuberous sclerosis
  - Children born with port-wine stain on upper face have a 15-50% chance of developing Sturge-Weber Syndrome
- Genetics
  - Activating mutation in GNAQ
  - Sporadic with somatic mosaicism
- Diagnosis is based off of presence of both the port wine stain and neuroimaging showing leptomeningeal capillary-venous malformations
  - Negative brain MRI at 1 year old can largely exclude disease

STURGE-WEBER SYNDROME

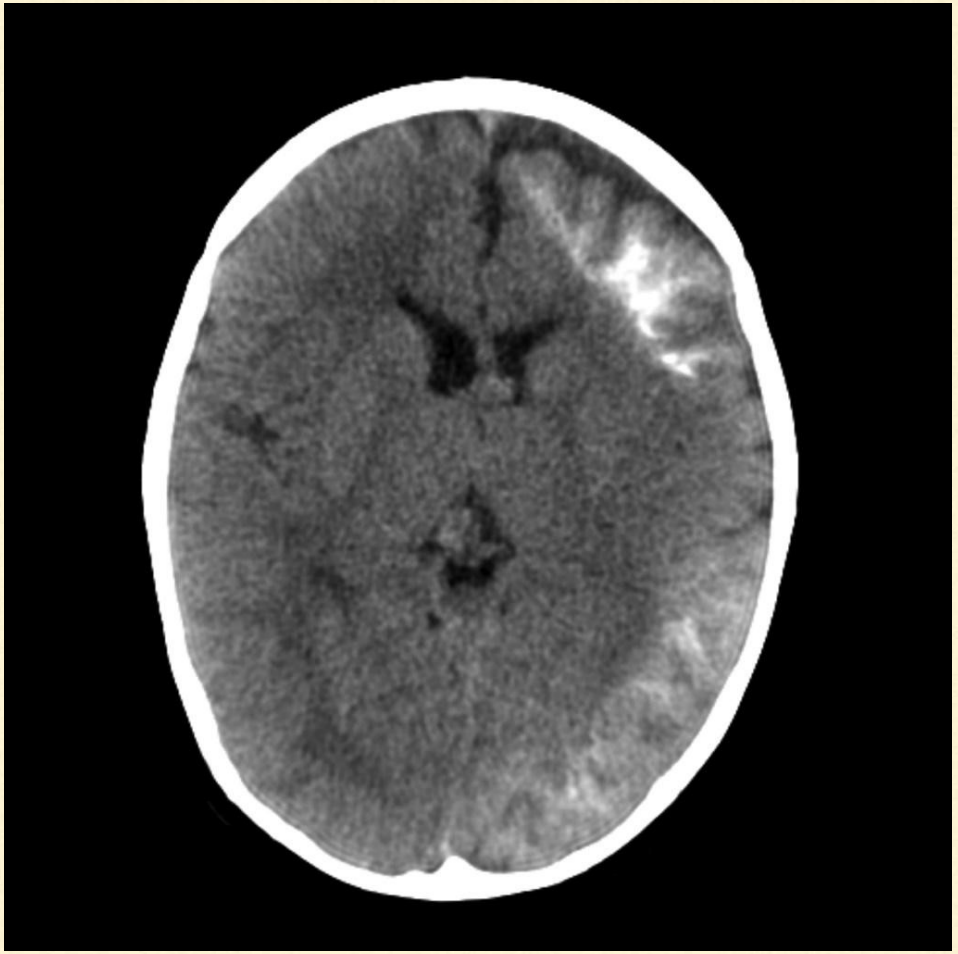
- Clinical Features: port wine stain (V1 & V2), seizures (80%), hemiparesis, intellectual disability (60%), homonymous hemianopia, glaucoma (30-70%)
- Management & Prognosis
  - Anti-epileptic therapy (carbamazepine vs surgery)
  - Low-dose aspirin
  - Prognosis correlates with degree of port wine stain and seizures



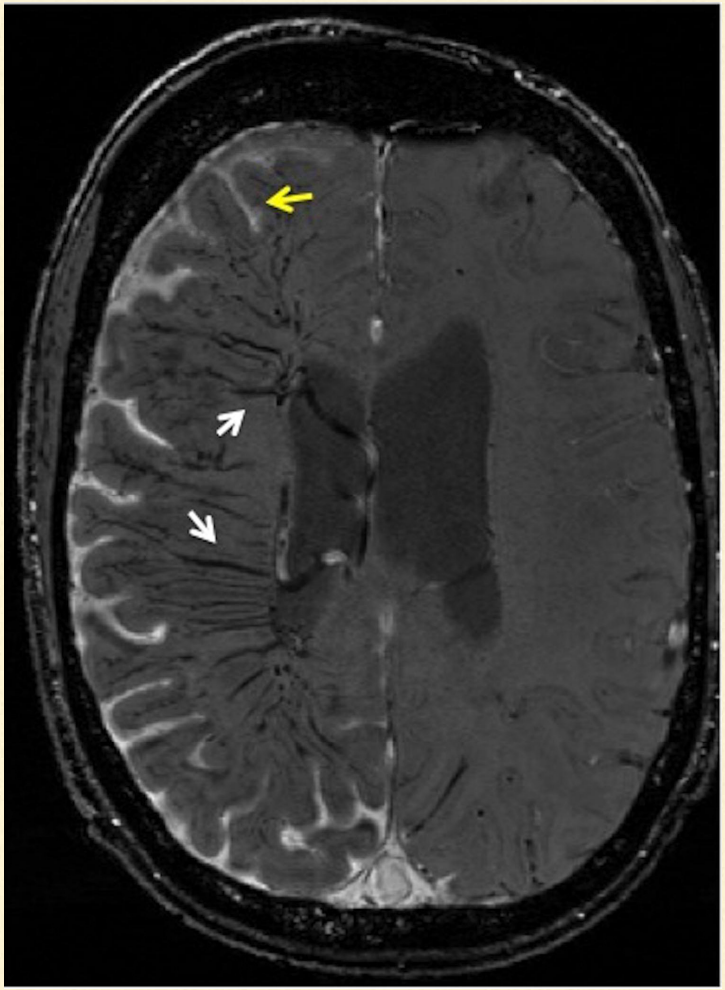
MRI FINDINGS

- Leptomeningeal enhancement from congestion of internal cerebral veins which leads to venous congestive ischemia and thus infarction/obliteration of parenchyma
  - Dilation of transparenchymal veins can also be seen
- Enlarged ipsilateral choroid plexus
- Leptomeningeal capillary-venous malformation may burn out later in life and lose contrast enhancement
- Anatomic loss of volume at older age
- *CT better images gyral “tram track” calcifications and also shows at an earlier age

LEPTOMENINGEAL CAPILLARY-VENOUS MALFORMATION



DILATION OF TRANSPARENCHYMAL VEINS



ACKNOWLEDGEMENTS

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